



## LCAT gene

lecithin-cholesterol acyltransferase

### Normal Function

The *LCAT* gene provides instructions for making an enzyme called lecithin-cholesterol acyltransferase (LCAT). This enzyme plays a role in removing cholesterol from the blood and tissues. Cholesterol is a waxy, fat-like substance that is produced in the body and obtained from foods that come from animals (particularly egg yolks, meat, poultry, fish, and dairy products). The body needs this substance to build cell membranes, make certain hormones, and produce compounds that aid in fat digestion. Too much cholesterol, however, increases a person's risk of developing heart disease, and can also lead to buildup of cholesterol in other tissues.

The LCAT enzyme helps transport cholesterol out of the blood and tissues by a process called cholesterol esterification. This process results in a form of cholesterol that is more efficiently carried by molecules called lipoproteins, which transport the cholesterol to the liver. Once in the liver, the cholesterol is redistributed to other tissues or removed from the body. The enzyme has two major functions, called alpha- and beta-LCAT activity. Alpha-LCAT activity helps attach cholesterol to a lipoprotein called high-density lipoprotein (HDL). Beta-LCAT activity helps attach cholesterol to other lipoproteins called very low-density lipoprotein (VLDL) and low-density lipoprotein (LDL).

### Health Conditions Related to Genetic Changes

#### complete LCAT deficiency

More than 70 mutations in the *LCAT* gene have been identified in people with complete LCAT deficiency, a disorder that primarily causes corneal clouding and kidney problems. Individuals with complete LCAT deficiency have mutations in both copies of the *LCAT* gene in each cell. Most of these mutations change single amino acids in the LCAT enzyme sequence. Others add or delete small amounts of genetic material in the *LCAT* gene. The mutations either prevent the production of LCAT or impair both alpha-LCAT and beta-LCAT activity, reducing the enzyme's ability to attach cholesterol to lipoproteins. Impairment of this mechanism for reducing cholesterol in the body leads to cholesterol deposits in the corneas, kidneys, and other tissues and organs. These deposits cause the signs and symptoms of complete LCAT deficiency.

## fish-eye disease

At least 18 mutations in the *LCAT* gene have been identified in people with fish-eye disease, also called partial LCAT deficiency. This disorder causes clouding of the clear covering of the eyes (corneas). Individuals with fish-eye disease have mutations in both copies of the *LCAT* gene in each cell. Most of these mutations change single protein building blocks (amino acids) in the LCAT sequence. The mutations impair alpha-LCAT activity, reducing the enzyme's ability to attach cholesterol to HDL. Impairment of this mechanism for reducing cholesterol in the body leads to cholesterol deposits in the corneas that gradually cause them to become cloudy.

## other disorders

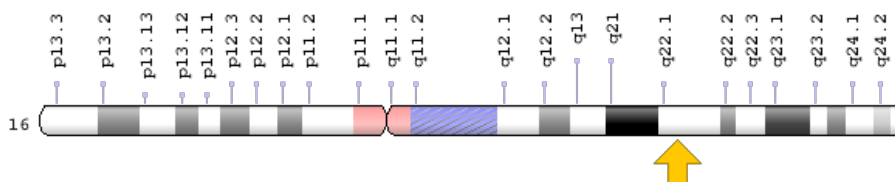
People with one *LCAT* gene mutation in each cell may have an increased risk of atherosclerosis, which is an accumulation of cholesterol-rich fatty deposits and scar-like tissue in the lining of the arteries that can impede blood flow and lead to heart attacks, strokes, and other health problems. A single *LCAT* gene mutation likely reduces alpha-LCAT activity and binding of cholesterol to HDL, leading to less efficient transport of cholesterol from the blood and resulting in the development of atherosclerosis.

Atherosclerosis also occurs in some people with fish-eye disease or complete LCAT deficiency. However, because atherosclerosis is very common in the general population and both fish-eye disease and complete LCAT deficiency are very rare, it is difficult to determine whether affected individuals are at increased risk of atherosclerosis.

## **Chromosomal Location**

Cytogenetic Location: 16q22.1, which is the long (q) arm of chromosome 16 at position 22.1

Molecular Location: base pairs 67,939,884 to 67,944,112 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- LCAT\_HUMAN
- phosphatidylcholine-sterol acyltransferase
- phosphatidylcholine-sterol acyltransferase precursor
- phospholipid-cholesterol acyltransferase

## Additional Information & Resources

### Educational Resources

- Madame Curie Bioscience Database: Fat Absorption and Lipid Metabolism in Cholestasis  
<https://www.ncbi.nlm.nih.gov/books/NBK6420/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28LCAT%5BTIAB%5D%29+OR+%28lecithin-cholesterol+acyltransferase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

### OMIM

- ATHEROSCLEROSIS SUSCEPTIBILITY  
<http://omim.org/entry/108725>
- LECITHIN:CHOLESTEROL ACYLTRANSFERASE  
<http://omim.org/entry/606967>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_LCAT.html](http://atlasgeneticsoncology.org/Genes/GC_LCAT.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=LCAT%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=6522](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6522)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/3931>
- UniProt  
<http://www.uniprot.org/uniprot/P04180>

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